



APOE gene

apolipoprotein E

Normal Function

The *APOE* gene provides instructions for making a protein called apolipoprotein E. This protein combines with fats (lipids) in the body to form molecules called lipoproteins. Lipoproteins are responsible for packaging cholesterol and other fats and carrying them through the bloodstream. Maintaining normal levels of cholesterol is essential for the prevention of disorders that affect the heart and blood vessels (cardiovascular diseases), including heart attack and stroke.

There are at least three slightly different versions (alleles) of the *APOE* gene. The major alleles are called e2, e3, and e4. The most common allele is e3, which is found in more than half of the general population.

Health Conditions Related to Genetic Changes

age-related macular degeneration

Alzheimer disease

The e4 version of the *APOE* gene increases an individual's risk for developing late-onset Alzheimer disease. People who inherit one copy of the *APOE* e4 allele have an increased chance of developing the disease; those who inherit two copies of the allele are at even greater risk. The *APOE* e4 allele may also be associated with an earlier onset of memory loss and other symptoms.

It is not known how the *APOE* e4 allele is related to the risk of Alzheimer disease. However, researchers have found that this allele is associated with an increased number of protein clumps, called amyloid plaques, in the brain tissue of affected people. A buildup of toxic amyloid beta peptide and amyloid plaques may lead to the death of neurons and the progressive signs and symptoms of this disorder.

It is important to note that people with the *APOE* e4 allele inherit an increased risk of developing Alzheimer disease, not the disease itself. Not all people with Alzheimer disease have the *APOE* e4 allele, and not all people who have this allele will develop the disease.

other disorders

Variants of apolipoprotein E have been studied extensively as risk factors for many different conditions. For example, *APOE* alleles have been shown to influence the

risk of cardiovascular diseases. People who carry at least one copy of the *APOE* e4 allele have an increased chance of developing atherosclerosis, which is an accumulation of fatty deposits and scar-like tissue in the lining of the arteries. This progressive narrowing of the arteries increases the risk of heart attack and stroke.

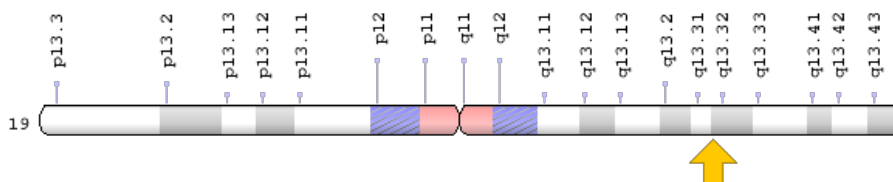
The *APOE* e2 allele has been shown to greatly increase the risk of a rare condition called hyperlipoproteinemia type III. Most people with this disorder have two copies of the *APOE* e2 allele, leading researchers to conclude that the e2 allele plays a critical role in the development of the condition. Hyperlipoproteinemia type III is characterized by increased blood levels of cholesterol, certain fats called triglycerides, and molecules called beta-very low-density lipoproteins (beta-VLDLs), which carry cholesterol and lipoproteins in the bloodstream. A buildup of cholesterol and other fatty materials can lead to the formation of small, yellow skin growths called xanthomas and the development of atherosclerosis.

APOE gene variants have also been studied as a potential risk factor for age-related macular degeneration, an eye disease that is a leading cause of vision loss among older people worldwide. Some studies have suggested that having at least one copy of the *APOE* e4 allele may help protect against this disease or delay the onset of vision loss, while having at least one copy of the *APOE* e2 allele may increase the risk of this disease or cause symptoms to appear earlier. However, other studies have not found these associations. More research is needed to clarify what role, if any, *APOE* gene variants play in the development of age-related macular degeneration.

Chromosomal Location

Cytogenetic Location: 19q13.32, which is the long (q) arm of chromosome 19 at position 13.32

Molecular Location: base pairs 44,905,749 to 44,909,395 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Apo-E
- APOE_HUMAN
- Apolipoproteins E

Additional Information & Resources

Educational Resources

- Eureka Bioscience Collection: Apolipoprotein E
<https://www.ncbi.nlm.nih.gov/books/NBK6203/#A15241>

GeneReviews

- Alzheimer Disease Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1161>
- APOE p.Leu167del-Related Lipid Disorders
<https://www.ncbi.nlm.nih.gov/books/NBK208534>

Genetic Testing Registry

- GTR: Genetic tests for APOE
<https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=348%5Bgeneid%5D>

Scientific articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28APOE%5BTI%5D%29+OR+%28apolipoprotein+E%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+genetics%5Bmh%5D+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D>

OMIM

- APOLIPOPROTEIN E
<http://omim.org/entry/107741>

Research Resources

- Alzheimer Research Forum: AlzGene database
<http://www.alzgene.org/geneoverview.asp?geneid=85>
- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_APOE.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=APOE%5Bgene%5D>

- HGNC Gene Family: Apolipoproteins
<http://www.genenames.org/cgi-bin/genefamilies/set/405>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=613
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/348>
- UniProt
<http://www.uniprot.org/uniprot/P02649>

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